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CHROMOSOMAL MICROARRAY ANALYSIS REPORT

Full Name / Ref No	**Fetus Of Roopam Verma**	Order ID/Sample ID	**630578/7933515**
Date of Birth / Age	**-**	Gender	**NK**
Parental Sample ID	7933523	Sample Type	**Amniotic fluid**
Referring Clinician	**Dr. Deepak Bansal** **Max Healthcare Institute Limited - New Delhi** **(New Delhi)**	Date and time of Sample Collection	**11-04-2023, 13:31:00**
		Date and time of Sample Receipt	**13-04-2023, 01:37:00**
		Date and time of Report	**22-04-2023, 07:13:56**
Test Requested	**Chromosomal Microarray - Affymetrix Cytoscan Optima low resolution genechip + Cell culture (If needed) [MGM1619]**		

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Fetus of Mrs. Roopam Verma, on antenatal scan revealed aberrant right subclavian artery, echogenic intracardiac foci in the left ventricle, and high risk for Trisomy 21 (1:39). The amniotic fluid sample is being evaluated for pathogenic Copy Number Variations (CNVs) by microarray analysis.

ARRAY TYPE

Affymetrix CytoScan™ Optima Array

ISCN NOMENCLATURE

CNVs	**46,**** (Normal copy number)
ROHs	-

RESULTS

CNVs: NO SIGNIFICANT COPY NUMBER VARIATION DETECTED

KARYOVIEW_CNVS

Note : As per Pre-Conception and Pre-Natal Diagnostic Testing (PCPNDT) Act 1994, the sex chromosome for this sample is masked.

INTERPRETATION

The cytogenomic microarray analysis indicated no clinically significant abnormalities and is consistent with a normal chromosome complement.

Note - Maternal Cell Contamination test result: 'Negative'.

Test result of most common anomalies

CONTENTS	RESULT
Autosomal Aneuploidies	
Trisomy 21 (Down syndrome)	Negative
Trisomy 18 (Edwards syndrome)	Negative
Trisomy 13 (Patau syndrome)	Negative
Other autosomal aneuploidies	Negative
Sex Chromosome Aneuploidies	
Monosomy X (Turner syndrome)	Negative
XYY (Jacobs syndrome)	Negative
XXY (Klinefelter syndrome)	Negative
XXX (Triple X syndrome)	Negative
Triploidy	Negative
Clinically significant Genome-wide copy number variations	
Gains	Negative
Losses	Negative

Microdeletions	
22q11 deletion (associated with DiGeorge Syndrome)	Negative
15q11 deletion (associated with Prader-Willi/ Angelman syndrome)	Negative
11q23 deletion (associated with Jacobsen syndrome)	Negative
8q24 deletion (associated with Langer-Giedion syndrome)	Negative
5p15 deletion (associated with Cri-du-chat syndrome)	Negative
4p16 deletion (associated with Wolf-Hirschhorn syndrome)	Negative
1p36 deletion syndrome	Negative
Other microdeletion / microduplication syndromes	Negative

REGIONS OF HOMOZYGOSITY

No significant regions of homozygosity changes or copy neutral long continuous stretches of homozygosity were detected.

RECOMMENDATIONS

Genetic counselling and additional testing may be warranted based on specific phenotypic indications.

TEST METHODOLOGY

Chromosomal microarray analysis (CMA) was performed using an Affymetrix CytoScan™ Optima array. This microarray consists of 315K oligonucleotide probes across the genome, including 18K unique non-polymorphic probes, and 148K bi-allelic SNP (single nucleotide polymorphism) probes. Genomic DNA (250 ng) was digested with Nsp1 and then ligated by Nsp1 adapter. Cytoscan Taq amplified PCR products of size 150 to 2200bp were purified using AMP pure beads and fragmented to the product size of 25bp to 125bp, biotin labelled, hybridized on CytoScan™ Optima gene chip, and then scanned. Data was analyzed using Chromosome Analysis Suite (ChAS) version 4.3.0.71. The analysis is based on the Human reference genome (GRCh38/hg38).

POSITIVE EVALUATION CRITERIA

Deletions smaller than 200 kb and duplications smaller than 500 kb may not be reviewed. Detected copy number variations (CNVs) are reported when found to have clear or suspected clinical relevance; CNVs devoid of relevant gene content or reported as common findings in the general population may not be reported. Regions of homozygosity are reported when a single LCSH is greater than 8-15 Mb (dependent upon chromosomal location and likelihood of imprinting disorder), or when the total autosomal LCSH proportion is greater than 3% (only autosomal LCSH greater than 3 Mb are considered for this estimate). Genomic linear positions are given relative to NCBI build 38 (hg38).

Test results are interpreted based on the recommendations and guidelines of International Standard of Cytogenomics Arrays (ISCA) as described below

Copy Number Change	A change in a segment of DNA at least 1kb in size that differ in copy number compared to reference genome. This could be either increase (Gain) or decrease (Loss) in chromosome number.
Pathogenic	This category includes CNVs, which overlaps with clearly established clinical significance. This usually means that a suspected disorder for which testing had been requested has been confirmed.
Likely Pathogenic	This category includes CNVs, that overlaps with a genomic region consistent with a syndrome containing OMIM morbid genes as well as deletions that overlap autosomal recessive genes (which may unmask a recessive allele associated with a syndrome/disorder).
Variants of unknown significance	This category includes CNVs, within a region which is not associated with genetic syndromes or symptoms of disease, deletions that overlap autosomal recessive genes (which may unmask a recessive allele but is not associated with a syndrome/disorder), de novo CNVs with no OMIM genes or genes associated with diseases
Likely Benign	The CNVs overlaps with the genome listed as benign in ISCA or other database based on large patient samples. Heterozygous duplication with no known OMIM morbid genes.
Benign	This category includes CNVs which are known not to be responsible for disease. Generally, no further action is warranted on such detections.

DISCLAIMER

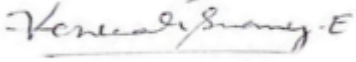
- This technique only identifies copy number variations viz., gain and losses along with regions of heterozygosity.
- Any other forms of polyploidy, truly balanced chromosome rearrangements (e.g., Inversions and balanced chromosomal imbalances), point mutation, small deletions and some mosaic conditions will not be detected.
- In accordance to the Pre-Conception and Pre-Natal Diagnostic Testing (PCPNDT) Act, 1994-Govt. of India; MedGenome Labs Ltd. does not disclose the gender of the fetus.

Dr. Satish Kumar K

Senior Scientist – Reproductive Genomics

Dr.Priya Kadam, MBBS, MSc.

Director - Reproductive Genomics



Dr. E. Venkataswamy, PhD

Lab Director

REFERENCES

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ABOUT THIS TEST:

Prenatal Aneuploidy Test evaluates genetic information in the sample (amniotic fluid/CVS/POC) provided, to detect aneuploidy of chromosomes 13, 18, 21, X or Y. This test does not detect other chromosomal or structural anomalies. Low level mosaicism involving chromosomes 13, 18, 21 X or Y may not be detected by this procedure.

PRENATAL ANEUPLOIDY TEST REPORT			
Patient Name	Fetus Of Roopam Verma	Order ID/Sample ID	630581/7933515
Gender	NK	Sample Type	Amniotic fluid
Maternal Age	26 Years	Date and Time of Samples Receipt	13.04.2023 01:37
Mother's Sample ID	7933523	Order Booked Date and Time	13.04.2023 13:09
Referring Clinician	Dr. Deepak Bansal	Date of report Generation	**15.04.2023**

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

The subject is being tested for common chromosomal abnormalities by QF-PCR and Microarray analysis. The antenatal findings revealed aberrant right subclavian artery, echogenic intracardiac foci in the left ventricle, and high risk for Trisomy 21 (1:39).

REPORT SUMMARY

No aneuploidy detected in Chromosomes 21, 18, 13 and sex chromosomes

Condition Tested	Result
TRISOMY 21	No aneuploidy detected.
TRISOMY 18	No aneuploidy detected.
TRISOMY 13	No aneuploidy detected.
Sex Chromosomes	No aneuploidy detected.

Interpretation:

Results are consistent with two copies of chromosomes 21,18, 13 and two sex chromosomes. The Amniotic fluid is not contaminated with maternal cells/tissues.

Testing method:

DNA is isolated from amniotic fluid/CVS/POC and using Devyser Compact v3 kit the markers specific to the chromosomes 13, 18, 21, X and Y are amplified. The amplified markers are then subjected to capillary electrophoresis. The data is analysed using GeneMapper™ (Thermofisher) and interpreted to determine aneuploidy of fetus for specific chromosomes.

Limitations:

1. This test is designed to detect for chromosome aneuploidies for chromosomes 21, 18, 13, X and Y.
2. These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal or sub chromosomal abnormalities, birth defects, and other conditions.
3. Low level mosaicism involving chromosomes 13, 18, 21 X or Y may not be detected by this test.
4. In accordance to the Pre-Conception and Pre-Natal Diagnostic Testing (PCPNDT) Act, 1994- Govt. of India; MedGenome Labs Ltd. does not disclose the gender of the fetus

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MedGenome Labs Pvt. Ltd. is registered in Bengaluru under the Prenatal Diagnostic Techniques (Regulation and Prevention of Misuse) Act,1994 Registration No. 1451

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